



## sialuria

Sialuria is a rare disorder that has variable effects on development. Affected infants are often born with a yellow tint to the skin and the whites of the eyes (neonatal jaundice), an enlarged liver and spleen (hepatosplenomegaly), and unusually small red blood cells (microcytic anemia). They may develop a somewhat flat face and distinctive-looking facial features that are described as "coarse." Temporarily delayed development and weak muscle tone (hypotonia) have also been reported.

Young children with sialuria tend to have frequent upper respiratory infections and episodes of dehydration and stomach upset (gastroenteritis). Older children may have seizures and learning difficulties. In some affected children, intellectual development is nearly normal.

The features of sialuria vary widely among affected people. Many of the problems associated with this disorder appear to improve with age, although little is known about the long-term effects of the disease. It is likely that some adults with sialuria never come to medical attention because they have very mild signs and symptoms or no health problems related to the condition.

### Frequency

Fewer than 10 people worldwide have been diagnosed with sialuria. There are probably more people with the disorder who have not been diagnosed, as sialuria can be difficult to detect because of its variable features.

### Genetic Changes

Mutations in the *GNE* gene cause sialuria. The *GNE* gene provides instructions for making an enzyme found in cells and tissues throughout the body. This enzyme is involved in a chemical pathway that produces sialic acid, which is a simple sugar that attaches to the ends of more complex molecules on the surface of cells. By modifying these molecules, sialic acid influences a wide variety of cellular functions including cell movement (migration), attachment of cells to one another (adhesion), signaling between cells, and inflammation.

The enzyme produced from the *GNE* gene is carefully controlled to ensure that cells produce an appropriate amount of sialic acid. A feedback system shuts off the enzyme when no more sialic acid is needed. The mutations responsible for sialuria disrupt this feedback mechanism, resulting in an overproduction of sialic acid. This simple sugar builds up within cells and is excreted in urine. Researchers are working to determine how an accumulation of sialic acid in the body interferes with normal development in people with sialuria.

## **Inheritance Pattern**

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most reported cases have occurred in people with no known history of the disorder in their family and may result from new mutations in the gene.

## **Other Names for This Condition**

- French type sialuria
- Sialuria, French type

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Sialuria  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342853/>

### Other Diagnosis and Management Resources

- GeneReview: Sialuria  
<https://www.ncbi.nlm.nih.gov/books/NBK1164>
- MedlinePlus Encyclopedia: Hepatosplenomegaly (image)  
<https://medlineplus.gov/ency/imagepages/17215.htm>
- MedlinePlus Encyclopedia: Newborn Jaundice  
<https://medlineplus.gov/ency/article/001559.htm>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Encyclopedia: Hepatosplenomegaly (image)  
<https://medlineplus.gov/ency/imagepages/17215.htm>
- Encyclopedia: Newborn Jaundice  
<https://medlineplus.gov/ency/article/001559.htm>
- Health Topic: Metabolic Disorders  
<https://medlineplus.gov/metabolicdisorders.html>

### Genetic and Rare Diseases Information Center

- Sialuria, French type  
<https://rarediseases.info.nih.gov/diseases/4865/sialuria-french-type>

### Educational Resources

- Cincinnati Children's Hospital Medical Center: Gastroenteritis  
<https://www.cincinnatichildrens.org/health/g/gastroenteritis>
- Disease InfoSearch: Sialuria, French type  
<http://www.diseaseinfosearch.org/Sialuria%2C+French+type/6588>
- KidsHealth from the Nemours Foundation: Anemia  
<http://kidshealth.org/en/parents/anemia.html>
- MalaCards: sialuria  
<http://www.malacards.org/card/sialuria>
- Orphanet: Sialuria  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=3166](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3166)

### Patient Support and Advocacy Resources

- Children Living with Inherited Metabolic Diseases (CLIMB) (UK)  
<http://www.climb.org.uk/>
- Resource list from the University of Kansas Medical Center: Metabolic Conditions  
<http://www.kumc.edu/gec/support/metaboli.html>

### GeneReviews

- Sialuria  
<https://www.ncbi.nlm.nih.gov/books/NBK1164>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28sialuria%5BTIAB%5D%29+OR+%28french+type+sialuria%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- SIALURIA  
<http://omim.org/entry/269921>

### **Sources for This Summary**

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